AI-DRIVEN METHODS FOR AUTOMATED ANALYSIS OF RETINAL DISEASES USING OPHTHALMIC IMAGING

SHORT SYNOPSIS

Submitted in partial fulfillment of the requirement of the degree of

DOCTOR OF PHILOSOPHY

by

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DECLARATION BY THE SCHOLAR

I hereby declare that the short synopsis entitled **AI-Driven Methods for Automated Analysis of Retinal Diseases Using Ophthalmic Imaging** being submitted in partial fulfillment of the requirement for the degree of Doctor of Philosophy in the Department of Computer Science & Technology under the School of Engineering of Manav Rachna University, Faridabad during the academic year 2025-2026

It is the work that is expected to be carried out under the supervision of **Dr. Dipali Bansal** and co-supervision of **Dr. Devanjali Relan**.

Further, it is declared that the short synopsis is within the plagiarism limit and copy of the same is attached as per the University norms.

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CERTIFICATE BY SUPERVISOR(S)

This is to certify that, the short synopsis entitled **AI-Driven Methods for Automated Analysis of Retinal Diseases Using Ophthalmic Imaging** is an original piece of work done by research scholar **Mr. Ashish Kumar Nayyar** under the supervision of **Dr. Dipali Bansal** and co-supervision of **Dr. Devanjali Relan** for the degree of Doctor of Philosophy in the Department of Computer Science & Technology under the School of Engineering of Manav Rachna University, Faridabad during the academic year 2025-2026.

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ABSTRACT

Retinal disease identification using fundus imaging is an important topic in medical field. Researchers are using deep learning models for early detection of diseases. These models are giving good results in detecting diseases like macular degeneration, diabetic retinopathy and glaucoma. Efficiency of a Deep learning model depends heavily of the dataset. The dataset showed be balanced and have lot of data sufficient enough for the models to learn. In real datasets like the RFMiD dataset, this is not the case. In these datasets some diseases have many images. But rare and severe diseases have very few samples. Because of this, the model cannot learn properly. It struggles to identify these rare conditions.

In this work, for automatic retinal disease detection, an AI-based system will be used. Convolutional neural network CNN will be used for classification whereas to solve the data imbalance problem, clinically guided Generative Adversarial Networks GANs will be used. GANs help in generating new but rare-disease focused synthetic fundus images. These synthetic fundus images generated by GAN will look similar to real ones. These synthetic images are mainly created for rare disease classes. They are then added to the original dataset to form an augmented dataset. Then the model will be trained using this new dataset. Its performance will be checked using accuracy, precision, recall, and F1-score. In addition to this clinical validations will also be done. This helps in understanding the improvement and medical relevance of the images.

Explainable AI will also be used in this work. Grad-CAM will be applied to the images. It highlights the important regions in the retina. This shows how the model is making decisions. It makes the system more understandable and useful for doctors. The main aim of this work is to improve AI detection performance. At the same time, the model should be easy to understand. The results will show that GAN-based augmentation is helpful. Explainable AI also adds value.

Keywords: Retinal fundus images, Retinal disease progression, Generative Adversarial Networks, Synthetic medical images, Deep learning, RFMiD dataset, Explainable AI.

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1. INTRODUCTION

Blindness and vision problems affect millions of people across the world. These problems reduce quality of life. They also create a burden on families and healthcare systems. In many cases, vision loss can be avoided. Proper screening and timely care can make a difference. Some eye diseases can be treated if they are detected at an early stage [1]. Delay in diagnosis often leads to permanent damage.

Retinal images are very important for eye disease analysis. Doctors use these images to examine the condition of the eye. The retina is a sensitive layer inside the eye. Many eye diseases show clear signs on the retina. Changes in blood vessels, lesions, and tissue damage can be observed in fundus images. Because of this, retinal imaging is widely used in ophthalmology.

However, analysing retinal images is not easy. It requires trained doctors and specialists. The process takes time and careful observation. Skilled experts are not always available. This problem is more serious in rural and remote areas. Many clinics do not have access to experienced ophthalmologists. In such cases, patients may not receive proper evaluation. It is also not possible for every hospital or research centre to consult experts regularly. This increases the workload on available specialists. As a result, diagnosis may be delayed. In some cases, patients may not receive treatment at the right time. This can lead to worsening of the disease.

In real clinical practice as most patients visit clinics only once or twice the datasets lacks repeated retinal scans of the same patient. Most patients do not go for a regular follow-up because of which repeated scans are not common. Because of this, it becomes difficult to collect multiple time-point image of same patient in the datasets. As a result most publicly available datasets contain retinal fundus images taken at a single time point.

These datasets are useful for detecting whether a disease is present and they only help in classification tasks. However, they do not provide information about how the disease progresses over time. These datasets thus cannot be directly used to study disease progression.

Longitudinal datasets, which contain images of the same patient over time, are not only very limited, they are also not openly available for research use.

Another major problem is of dataset imbalance in medical imaging. It happens when some classes in a dataset have many samples, while others have very few. In retinal datasets, this problem is very clear common diseases like diabetic retinopathy and glaucoma have more images. Glaucoma is a common eye disease. In Glaucoma the increased pressure inside the eye damages the end of optic nerve. This damage mainly affects the nerve fibers at the optic disc. This leads to gradual loss of side vision. If not treated early, it can result in permanent blindness. Another issue is that rare conditions have very limited data in datasets. Because of this, the model gets biased as it learns more from the disease classes that occur majorly. It starts ignoring the minority classes. This affects the overall performance of the system. Even if accuracy looks high, the model may fail to detect rare diseases. This problem becomes more serious in real-world applications where missing a rare disease can lead to serious consequences. Traditional methods like oversampling and under sampling are used, but they have limitations. Oversampling can lead to overfitting. Under sampling can result in loss of useful data. That is why advanced methods like data augmentation and GANs are used. These methods help in creating more balanced datasets. By improving data distribution, the model can learn better features. This leads to better detection of both common and rare diseases.

Due to this limitation, researchers face difficulty in studying disease identification and progression. This affects the development of predictive models. It also limits research on treatment planning and outcome analysis. To address this problem, a deep learning-based approach is proposed. The study uses Generative Adversarial Networks. It studies patterns related to disease appearance and applies augmentation techniques to generate synthetic images. This approach helps reduce the data gap & provides additional images for improved AI analysis. It supports better understanding of disease development. Doctors and researchers can use this data for early detection of rare disease. This is especially useful in areas with limited eye care facilities. With better monitoring tools, the risk of severe vision loss can be reduced. This can improve patient care and overall eye health outcomes.

2. LITERATURE REVIEW

Retinal fundus imaging is widely used in the analysis of eye diseases. It helps doctors observe internal structures of the eye. Diseases such as diabetic retinopathy, glaucoma, and age-related macular degeneration show clear signs on the retina.

Glaucoma is a progressive disease of the eye. It damages the optic nerve and affects vision. This happens due to loss of retinal ganglion cells. The damage is permanent and cannot be reversed. It is one of the major causes of blindness worldwide [2]. Many people are affected across different age groups. High intraocular pressure is a major risk factor. But glaucoma can also occur even when pressure is normal. This shows that the disease is complex in nature [3].

Recent studies try to explain how glaucoma develops at the molecular level. Increased eye pressure creates stress at the optic nerve head. This leads to damage of nerve fibers. It also causes changes in the surrounding tissue. Over time, retinal ganglion cells start dying [4]. Other factors like oxidative stress and inflammation also play a role. Blood flow problems in the eye are also linked with the disease [5]. Some studies have found genes related to glaucoma. These findings may help in early diagnosis and new treatments [6].

Treatment mainly focuses on reducing eye pressure. This helps to slow down the disease. But new research is looking beyond pressure control. Neuro-protection is being studied to save optic nerve cells. New methods like drug delivery using nanoparticles are being tested. Implants and gene therapy are also being explored [7]. Artificial intelligence is now used in glaucoma detection. It helps in early diagnosis and tracking disease progression [8].

Overall, glaucoma is not caused by a single factor. It is a complex disease with many causes. Better diagnosis and treatment methods are needed. Future research is focusing on combining genetics, biology, and AI tools. This may help in early detection and prevention of vision loss [9].

Because of this, retinal fundus images are commonly used for screening and diagnosis[10].

In recent years, deep learning techniques have been widely applied to retinal image analysis. Convolutional Neural Networks are the most commonly used models. These networks automatically learn features from images. Many studies have used CNNs for retinal disease detection and classification [11]. Some works also focus on grading disease severity[12],[13].

Most deep learning models are trained on cross-sectional datasets. These datasets contain only one image per patient. The images are captured at a single time point. Such data is useful for disease detection. However, it does not provide information about disease progression over time. Disease progression is a gradual process. Retinal diseases usually evolve from early stages to advanced stages. Existing datasets do not capture this temporal change. As a result, most models treat each retinal image independently. Temporal relationships between disease stages are ignored [14].

Several public retinal datasets are available. These include EyePACS, Messidor, REFUGE, DRIVE, STARE, and RFMiD. These datasets are widely used in research. They provide high-quality fundus images with disease labels. However, they lack longitudinal information. Images of the same patient across different time points are not available[15]

Many studies show that retinal image datasets are small and unbalanced. To address data scarcity, generative models have been introduced. Generative Adversarial Networks are widely used in medical imaging. GANs help by creating more data. GANs consist of a generator and a discriminator. The generator creates synthetic images. The discriminator checks image realism. Through adversarial training, GANs like WGAN-GP can generate realistic medical images[16].

In retinal imaging, GANs are mainly used for data augmentation. Synthetic images are generated to increase dataset size. GANs are also used for image enhancement tasks such as denoising and resolution improvement [17].

Image-to-image translation models such as Pix2Pix have been applied to retinal images. These models learn a mapping between two image domains using paired data. CycleGAN is used when paired data is not available. It has been applied for retinal image style transfer and modality translation [18], [19].

Although GAN-based methods improve visual quality, they do not focus on disease progression. Most synthetic images are not linked to clinical severity stages. The transition from mild to moderate or severe disease is not explicitly learned. The focus remains on visual realism rather than clinical correctness [20].

Some studies have attempted disease progression prediction using deep learning. These approaches predict future disease states from baseline images. In most cases, progression is modeled directly from normal to diseased states. Intermediate stages are often ignored. This does not reflect real clinical progression [21].

Not much work has been done on multi-stage and multi-disease progression in retinal conditions. Studying disease step by step, like mild to moderate or moderate to severe, can show real disease behavior better. Most studies look at only one disease. But in real life, patients can have more than one retinal condition. Very few methods handle both multiple diseases and multiple stages [22]. Some recent studies focus on classifying retinal blood vessels in fundus images. This helps find useful biomarkers for eye and systemic diseases. Mokan *et al.* made a model that classifies arteries and veins pixel by pixel using supervised learning [23]. Their work helps understand structural changes in the retina. It can also support multi-stage, multi-disease modeling.Another major limitation is evaluation. Most synthetic image studies rely on metrics such as SSIM, PSNR, and FID. These metrics measure visual similarity. They do not confirm whether disease progression is clinically meaningful. A visually realistic image may still be clinically incorrect [24].

Explainability is also lacking in many studies. Deep learning models often act as black boxes. Without interpretability, clinicians may not trust generated images. Few studies use explainable AI techniques to validate synthetic retinal images [25].

The RFMiD dataset provides retinal images for multiple diseases along with severity labels. This makes it suitable for multi-disease analysis [26]. However, it does not contain true longitudinal scans of the same patient. This highlights the need for synthetic longitudinal progression modeling.

From the literature, it is clear that most research focuses on common disease detection or image synthesis. Very limited work addresses rare retinal disease which are largely unexplored. Deep Learning techniques help in creating more balanced datasets and improved learning. This gap motivates the proposed research.

A few Rare Retinal Diseases

Central Retinal Artery Occlusion (CRAO):

CRAO is a vision-threatening condition caused by blockage of the central retinal artery, leading to sudden and painless loss of vision along with characteristic retinal whitening and a cherry-red spot [27].

Macular Hole (MH):

Macular Hole is a full-thickness defect in the macular region of the retina, resulting in loss of central vision due to disruption of retinal layers [28].

Optic Disc Edema (ODE):

Optic Disc Edema refers to swelling of the optic nerve head, often caused by increased intracranial pressure or inflammation, and appears as blurred disc margins in fundus images [29].

Branch Retinal Vein Occlusion (BRVO):

BRVO occurs due to blockage of a branch of the retinal vein, leading to localized retinal hemorrhages, edema, and vision impairment in the affected region [30].

Hypertensive Retinopathy (HR – Severe):

Severe hypertensive retinopathy is associated with high blood pressure and presents with retinal haemorrhages, cotton-wool spots, exudates, and in advanced cases, optic disc swelling [30].

3. RATIONALE/SIGNIFICANCE OF THE STUDY

This study is motivated by the limitations of current retinal disease detection methods. Most models require large and balanced datasets. In practice, such data is not available. Datasets like RFMiD include many disease categories, but some classes have very few samples. Because of this, models do not learn rare disease patterns well. This creates a mismatch between experimental results and real clinical use.

Another concern is the absence of longitudinal data. Most datasets provide images from a single visit. These images help in identifying disease, but they do not show how the condition changes over time. Without follow-up data, it is difficult to study disease progression. It also limits prediction of future severity. This reduces the usefulness of AI systems in long-term monitoring.

There is also a shortage of specialists in many regions. Retinal image analysis is a time-consuming task. It requires trained professionals. In many rural areas, such expertise is not available. As a result, diagnosis is often delayed. In some cases, patients are not screened at all. This increases the risk of avoidable vision loss. Automated systems can help reduce this gap.

Synthetic data generation provides a possible solution to this problem. By using deep learning models, new retinal images can be generated from existing data. These images can represent different disease stages. This helps in reducing the gap caused by the lack of longitudinal datasets.

Generative models offer a practical direction to handle data-related issues. GANs can create synthetic retinal images that look realistic. These images can be used to increase samples for underrepresented classes. This improves class balance in the dataset. It also helps models learn better features for rare diseases. At the same time, it reduces dependence on large-scale data collection.

The study aims to improve the reliability of automated retinal analysis. The study also considers clinical relevance and explainability. Visual quality alone is not sufficient in medical

applications. The generated images must also make sense from a clinical point of view. Explainable AI techniques help in understanding what the model learns. This improves trust and transparency.

Better data representation leads to better model performance. The use of synthetic data supports more stable training. It also improves detection across different disease categories. This can assist both researchers and clinicians. Early detection becomes more feasible. Overall, the work supports better diagnosis and may help reduce preventable blindness.

Overall, this study is significant for both research and healthcare. It supports the creation of useful synthetic datasets. It also promotes responsible use of artificial intelligence in medical imaging. The work can help improve future research, diagnosis, and disease monitoring.

4. PROBLEM STATEMENT

To develop AI-driven methods to automatically analyze ophthalmic images for detecting and assessing multiple retinal diseases, enabling faster, more accurate, and scalable diagnosis while addressing challenges like data imbalance and variability in disease severity.

5. RESEARCH OBJECTIVES

RO1: To develop deep learning models for automated retinal disease detection including glaucoma.

RO2: To investigate data-efficient learning techniques for limited ophthalmic datasets.

RO3: Develop generative models for synthetic retinal image generation and augmentation.

RO4: To explore explainable AI methods to improve model interpretability for clinicians.

RO5: To develop a clinical decision support framework for retinal disease screening.

6. RESEARCH METHODOLOGY

This research follows a structured methodology to address the challenges of data scarcity and class imbalance in retinal disease detection. Figure 2. Shows the methodology to be used in this research.

6.1 In addition to RFMiD, publicly available glaucoma datasets such as REFUGE [31] and DRISHTI-GS [32] will be utilized to enhance model generalization and evaluate disease-specific performance. The RFMiD dataset contains retinal fundus images annotated with multiple disease labels. The dataset includes approximately 35 different retinal disease categories, covering both common and rare conditions. However, a major limitation of this dataset is its imbalance. Some diseases are well represented with a large number of images, while others, especially rare and severe conditions, have very limited samples. This imbalance creates difficulty during model training, as deep learning models tend to perform poorly on underrepresented classes.

6.2 To address this issue, the main approach of the research focuses on reducing the effects of data scarcity and imbalance through synthetic data generation. In this work, each image is treated as an independent sample, and no relationship between images is assumed. There is no use of patient history or temporal progression data. The goal is to improve classification performance by enhancing the dataset rather than modelling disease progression over time.

6.3 For image generation, a conditional Generative Adversarial Network (GAN) is employed. The model takes disease labels as input and generates corresponding retinal fundus images. This allows the generation process to be controlled based on specific disease categories. Special emphasis is placed on generating images for rare and severe disease classes, where data is limited. The GAN learns the distribution of real images and produces synthetic images that resemble real fundus images in terms of structure and visual patterns.

6.4 The generated synthetic images are then used for dataset augmentation. These images are added only to the underrepresented classes to improve class balance. The augmented dataset is created by combining the synthetic images with the original dataset. This results in a more balanced training dataset, which helps the model learn better representations for minority classes. It is important to note that the test dataset remains unchanged and consists only of real images to ensure a fair evaluation of model performance.

6.5 For model training and evaluation, a deep learning-based classification model is trained using the augmented dataset. The performance of the model is evaluated using metrics such as F1-score, recall, and balanced accuracy. These metrics are particularly useful for imbalanced datasets, as they provide a more reliable assessment compared to simple accuracy. Special attention is given to the performance on rare disease classes to verify whether the augmentation strategy improves their detection. The results obtained from

the augmented dataset are then compared with those from a model trained on the original dataset to measure the effectiveness of the proposed approach.

6.6 Finally, the experimental setup involves multiple stages. First, the quality of the generated synthetic images is evaluated using the Fréchet Inception Distance (FID) score, which measures how close the synthetic images are to real images. After this, two main experiments are conducted: one using the original dataset and the other using the augmented dataset. The results from both experiments are compared to analyze the improvement achieved through synthetic data generation and augmentation. This comparison helps in validating the effectiveness of the proposed methodology in handling data imbalance and improving model performance.

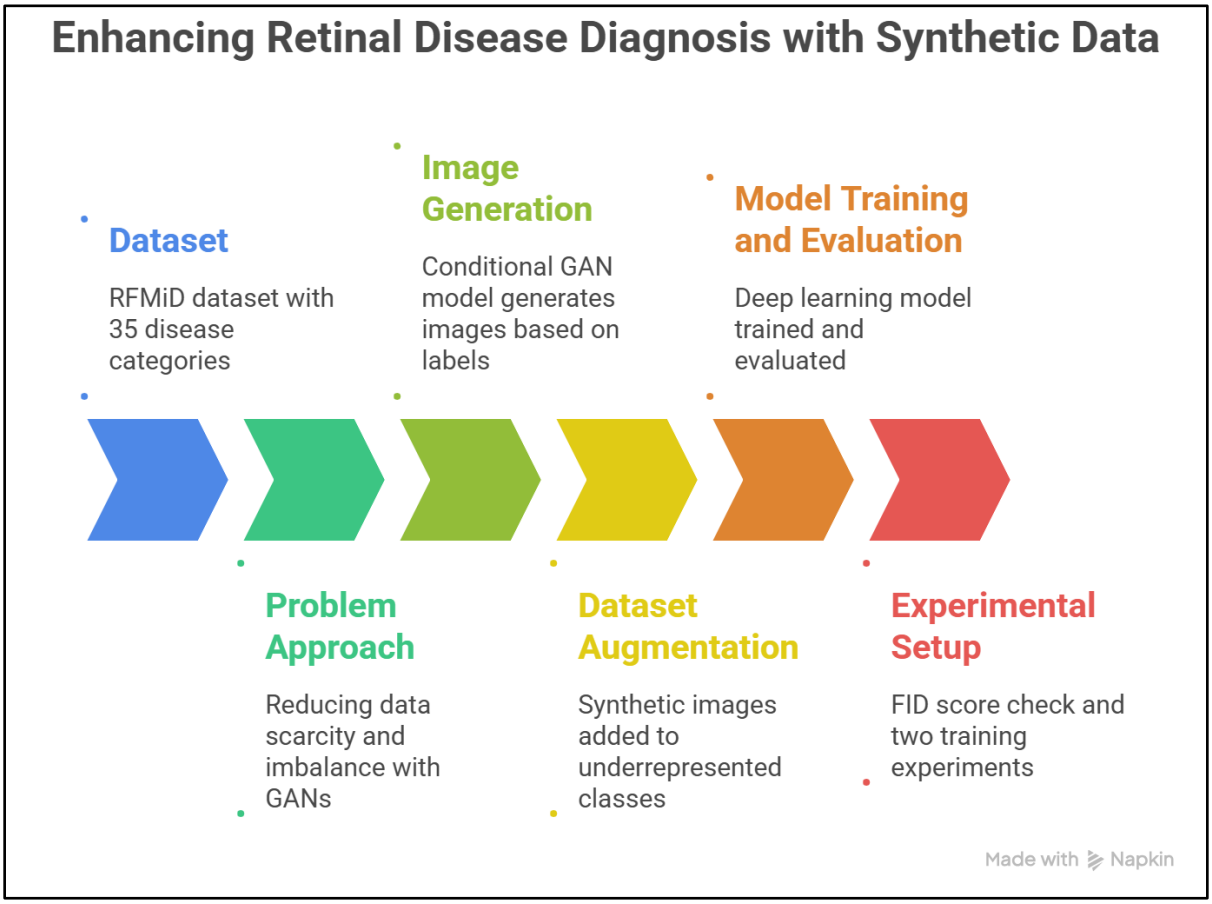
Enhancing Retinal Disease Diagnosis with Synthetic Data
Dataset
RFMiD dataset with 35 disease categories
Problem Approach
Reducing data scarcity and imbalance with GANs
Image Generation
Conditional GAN model generates images based on labels
Dataset Augmentation
Synthetic images added to underrepresented classes
Model Training and Evaluation
Deep learning model trained and evaluated
Experimental Setup
FID score check and two training experiments
Made with Napkin

Figure 1. Methodology to be used for Automated Analysis of Retinal Diseases Using Ophthalmic Imaging
Source: Napkin AI

Hardware & Software Requirements

Software Requirements

This research work is mainly implemented using cloud-based platforms and Python-based tools. The primarily **Kaggle platform** is used for development, Kaggle provides an integrated environment for coding, dataset management, and model training. It offers free access to GPU resources, which makes it suitable for training deep learning models such as Generative Adversarial Networks (GANs). It also allows easy access to publicly available datasets, including retinal image datasets.

For additional experimentation and flexibility, **Jupyter Notebook** is used for local testing, while **Google Colab** may be used as an alternative cloud environment when needed.

The main software components used for data preprocessing, model development, training, evaluation, and visualization in this research include:

- **Programming Language:** Python (3.x)
- **Deep Learning Frameworks:** PyTorch / TensorFlow
- **Libraries:** NumPy, Pandas, OpenCV, Matplotlib, Scikit-learn
- **Development Platforms:** Kaggle (primary), Jupyter Notebook, Google Colab

Hardware Requirements

Kaggle's cloud-based computational resources with the following approximate configuration were used:

- **GPU:** NVIDIA Tesla T4 / P100 (varies by session availability)
- **RAM:** Approximately 13–16 GB
- **Storage:** Temporary working storage (~20 GB)

In addition to above following configuration was used for local work (such as code editing, lightweight preprocessing, and result analysis):

- **CPU:** Intel Core i5
- **RAM:** 8 GB
- **Storage:** 256 GB SSD and 1TB HDD

7. EXPECTED OUTCOME OF THE RESEARCH

The main outcome of this work is improvement in retinal disease detection including structural disease like glaucoma using the publicly available datasets, which are highly imbalanced. This research is an attempt to make the model more reliable even for datasets with limited data for rare diseases.

It is expected that GAN-based image augmentation will increase the number of samples for rare disease classes. The generated synthetic images should appear to look close to real fundus images and show proper disease features. This can help in reducing the imbalance problem to some extent. After adding synthetic images, the dataset becomes more balanced. This approach shall improve models learning especially in rare and severe disease categories. The improvement is expected to be visible in F1-score, recall, and balanced accuracy.

The model trained on the augmented dataset shall perform better than the one trained on the original dataset. This comparison will show whether synthetic data is actually useful or not. The quality of generated images is also important. The GAN model should produce images with a low FID score. This will indicate that the generated images are close to real data and not random noise.

Another outcome is related to learning with limited data. The research should show that even with small and imbalanced datasets, good performance can be achieved using augmentation methods. This can be useful in medical imaging where data collection is difficult.

In the end, the work should help in developing better AI-based retinal disease detection systems. Such systems can support doctors in early diagnosis, especially in rare cases where data is usually not available.

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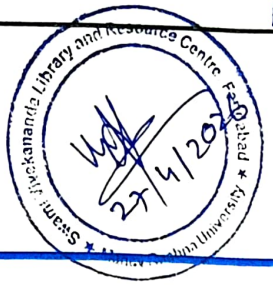

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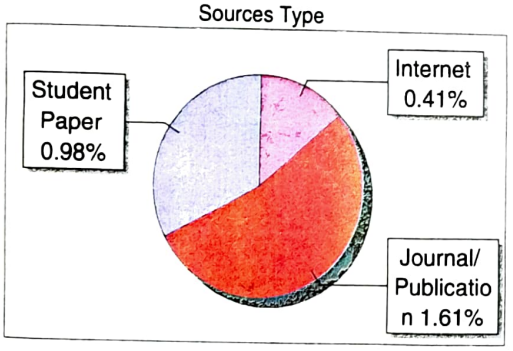

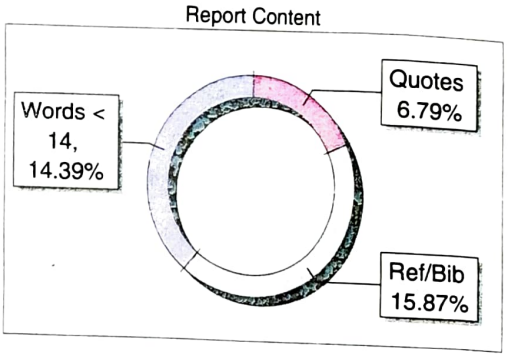

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Ashish Nayyar Dipali

Annexure-6

AI-DRIVEN METHODS FOR AUTOMATED ANALYSIS OF RETINAL DISEASES USING OPHTHALMIC IMAGING

SHORT SYNOPSIS

by

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Under the supervision of

Dr. Dipali Bansal
Professor

Dr. Devanjali Relan
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Year - 2026

ABSTRACT

Retinal disease identification using fundus imaging is an important topic in medical field. Scholars are using deep learning models for detection of diseases. These models presents good results in detecting diseases like glaucoma, macular degeneration and diabetic retinopathy. The dataset should be balanced. It should also have enough data for proper learning. In real datasets like RFMiD, this is not true as common diseases have many images. Problem is that some rare and serious diseases have very few image samples. Because of this, the model does not learn well. It becomes tough to detect rare diseases. In this work, an deep learning model are used for disease classification purpose. To handle the imbalance problem, Generative Adversarial Networks (GANs) are used with clinical guidance. GANs help in generating new but rare-disease focused synthetic fundus images. These synthetic fundus images generated by GAN will look similar to real ones. These synthetic images are mainly created for rare disease classes. They are then added to the original dataset to form an augmented dataset. Then the model will be trained using this new dataset. Its performance will be checked using accuracy, precision, recall, and F1-score. In addition to this clinical validations will also be done. This helps in understanding the improvement and medical relevance of the images.

Explainable AI will also be used in this work. Grad-CAM will be applied to the images. It highlights the important regions in the retina. This shows how the model is making decisions. It makes the system more understandable and useful for doctors. The main aim of this work is to improve AI detection performance. At the same time, the model should be easy to understand. The results will show that GAN-based augmentation is helpful. Explainable AI also adds value.

Keywords: Retinal fundus images, Retinal disease progression, Generative Adversarial Networks, Synthetic medical images, Deep learning, RFMiD dataset, Explainable AI.

1. INTRODUCTION

Blindness and vision problems affect millions of people across the world. These problems reduce quality of life. They also create a burden on families and healthcare systems. In many

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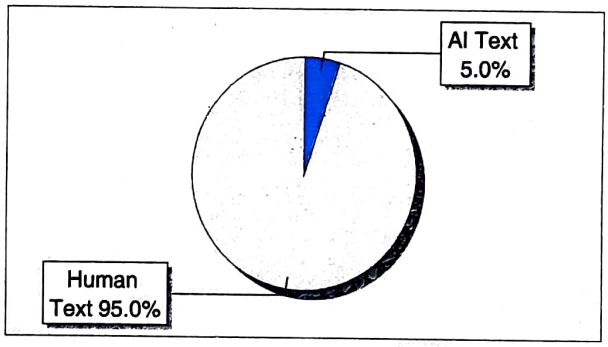

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